GRHPR gene

glyoxylate and hydroxypyruvate reductase

Normal Function

The *GRHPR* gene provides instructions for making an enzyme called glyoxylate and hydroxypyruvate reductase. This enzyme plays a role in preventing the buildup of a potentially harmful substance called glyoxylate by converting it to a substance called glycolate, which is easily excreted from the body. Additionally, this enzyme can convert a compound called hydroxypyruvate to D-glycerate, which is eventually converted to the simple sugar glucose (by other enzymes) and used for energy.

Health Conditions Related to Genetic Changes

primary hyperoxaluria

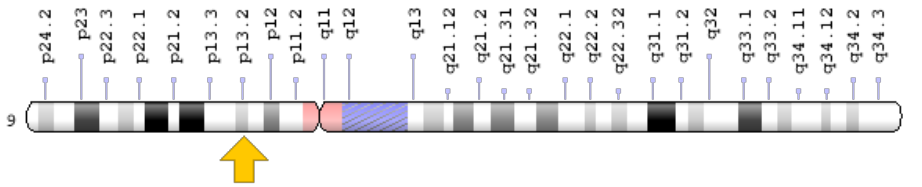
More than 25 mutations in the *GRHPR* gene have been found to cause primary hyperoxaluria type 2. This condition is caused by the overproduction of a substance called oxalate. Excess amounts of this substance lead to kidney and bladder stones, which begin in childhood and often result in kidney disease by early adulthood. Deposition of oxalate in multiple other tissues throughout the body (systemic oxalosis) can cause additional health problems.

GRHPR gene mutations either disrupt production of the glyoxylate and hydroxypyruvate reductase enzyme or alter its structure. As a result, enzyme activity is absent or severely reduced and the conversion of glyoxylate to glycolate is impaired. Glyoxylate builds up and is converted to a compound called oxalate. The oxalate is filtered through the kidneys and is either excreted in urine as a waste product or combines with calcium to form calcium oxalate, a hard compound that is the main component of kidney and bladder stones. Increased oxalate levels in the blood can lead to systemic oxalosis, particularly affecting bones and the walls of blood vessels in people with primary hyperoxaluria type 2.

Chromosomal Location

Cytogenetic Location: 9p13.2, which is the short (p) arm of chromosome 9 at position 13.2

Molecular Location: base pairs 37,422,666 to 37,437,782 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- D-glycerate dehydrogenase
- GLXR
- glyoxylate reductase/hydroxypyruvate reductase
- GRHPR_HUMAN
- PH2

Additional Information & Resources

GeneReviews

- Primary Hyperoxaluria Type 2
  https://www.ncbi.nlm.nih.gov/books/NBK2692

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GRHPR%5BTIAB%5D%29+OR+%28glyoxylate+reductase/hydroxypyruvate+reductase%5BTIAB%5D%29%29+OR+%28GLXR%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- GLYOXYLATE REDUCTASE/HYDROXYPYRUVATE REDUCTASE
  http://omim.org/entry/604296

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GRHPR.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GRHPR%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4570
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/9380
- UniProt
  http://www.uniprot.org/uniprot/Q9UBQ7

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